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AHI1 Rabbit pAb

Catalog # AP97313

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	Q8N157
Other Accession	Q8N157
Predicted	Rat, Pig, Human, Mouse, Rabbit, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	137115
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human AHI1
Epitope Specificity	801-900/1196
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Contains 1 SH3 domain.Contains 7 WD repeats.
SUBUNIT	Part of the tectonic-like complex (also named B9complex). Interacts with MKS1 (By similarity). Interacts withNPHP1.
DISEASE	Defects in AHI1 are the cause of Joubert syndrome type 3(JBTS3) [MIM:608629]. JBTS is an autosomal recessive disorderpresenting with cerebellar ataxia, oculomotor apraxia, hypotonia,neonatal breathing abnormalities and psychomotor delay.Neuroradiologically, it is characterized by cerebellar vermianhypoplasia/aplasia, thickened and reoriented superior cerebellarpeduncles, and an abnormally large interpeduncular fossa, givingthe appearance of a molar tooth on transaxial slices (molar toothsign). Additional variable features include retinal dystrophy andrenal disease. JBTS3 shows minimal extra central nervous systeminvolvement and appears not to be associated with renaldysfunction.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Highly expressed in the most primitive normal hematopoietic cells. Expressed in brain, particularly in neurons that give rise to the crossing axons of the corticospinal tract and superior cerebellar peduncles. Expressed in kidney (renal collecting duct cells) (at protein level). Involvement in disease:Defects in AHI1 are the cause of Joubert syndrome type 3 (JBTS3) . JBTS is an autosomal recessive disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. JBTS3 shows minimal extra central nervous system involvement and appears not to be associated with renal dysfunction.

Additional Information

Gene ID	54806
Other Names	AHI-1; JBTS3; ORF1; dJ71N10.1; 1700015F03Rik; D10Bwg0629e; AHI1_HUMAN; AHI1; Abelson helper integration site 1 protein homolog (AHI-1); AHI1_MOUSE; Abelson helper integration site 1 protein (AHI-1); AHI1_RAT; Abelson helper integration site 1; Abelson helper integration site; Jouberin
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500,ELISA=1:5000-10000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	AHI1
Function	Involved in vesicle trafficking and required for ciliogenesis, formation of primary non-motile cilium, and recruitment of RAB8A to the basal body of primary cilium. Component of the tectonic-like complex, a complex localized at the transition zone of primary cilia and acting as a barrier that prevents diffusion of transmembrane proteins between the cilia and plasma membranes. Involved in neuronal differentiation. As a positive modulator of classical Wnt signaling, may play a crucial role in ciliary signaling during cerebellum embryonic development (PubMed:21623382).
Cellular Location	Cytoplasm, cytoskeleton, cilium basal body. Cell junction, adherens junction. Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriole {ECO:0000250|UniProtKB:Q8K3E5}. Note=In the retinal photoreceptor cell layer, localizes at the connecting cilium {ECO:0000250|UniProtKB:Q8K3E5}
Tissue Location	Highly expressed in the most primitive normal hematopoietic cells. Expressed in brain, particularly in neurons that give rise to the crossing axons of the corticospinal tract and superior cerebellar peduncles. Expressed in kidney (renal collecting duct cells) (at protein level).

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.